

Evaluation of hemoglobinopathy carrier rates in Bilecik hemoglobinopathy diagnosis centre

Hemoglobinopathy prevalence in Bilecik

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Abstract

Aim: Hemoglobinopathies are important public health problems with high mortality and morbidity rates. Additionally, they are amongst the most common inherited disorders in Turkey and can be preventable with the help of premarital screening programs. The aim of the present study was to analyse the hemoglobinopathy test results in individuals who applied to the hemoglobinopathy diagnosis centre in Bilecik/Turkey. Material and Method: In our study, hemoglobinopathy test results from January 2006 to April 2017 in Bilecik Hemoglobinopathy Diagnostic Centre were evaluated retrospectively. Blood samples from individuals who applied for pre-marriage counselling and general screening were collected and hemoglobinopathies (Hb A0, A1C, A2, F, C, D, E, and S) were screened by using High Performance Liquid Chromatography (HPLC). Results: A total of 23,676 people were included in the present study. Beta-thalassemia trait was detected in 1,317 (5.5%) and, HbA2 at a level over 3.5% was detected in 1,177 (5.0%). Additionally, elevated HbF levels were over 2% of individuals (324/23,676). The positivity of hemoglobin D and HbS traits were 0.06% (15 patients) and 0.01% (two patients) respectively. Discussion: In the present study the positive rate of HbA2 was 4.6%. When compared this finding was higher than the overall average rate in Turkey. The rate is similar to those in studies from Istanbul and the Mediterranean region, but it is higher than in studies from Bursa and Kocaeli. Our results indicated that premarital hemoglobinopathy screening should be continued for all partners planning to get married and for each individual in Bilecik who may potentially have a hemoglobinopathy trait.

Keywords

Hemoglobinopathy; Bilecik; Beta-Thalassemia Trait; Sickle Cell Anaemia

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Introduction

Hemoglobinopathies are considered an important public health problem because of significant mortality and morbidity. On a global scale, the prevalence of abnormal Hb gene carriage is estimated as 7%. Clinically significant hemoglobinopathies are observed in 300,000 to 500,000 children worldwide each year, with the majority (80%) in developing countries [1]. According to Turkish Public Health Institute guidelines in 2016, the trait frequency of beta thalassemia was 2.1%; there were about 1.3 million traits identified and about 4.500 patients [2]. In a study including sixteen provinces in Turkey, the trait frequency was reported as 4.3%. The highest prevalence of the beta-thalassemia trait (13.1%) was found in Antalya and Adana [3]. In general, hemoglobinopathies are defined in two different ways. Globin protein has a role in the synthesis disorders (thalassemia) or in structural changes (abnormal hemoglobins) in the polypeptide chains of the hemoglobin molecule. In our country, 42 abnormal hemoglobins are known and HbS, HbD, and HbE are most common [4]. In our country, regions at risk first started to control hemoglobinopathy. and Then, pre-marriage screening tests spread all over the country, preventing the birth of sick children. Preventive medicine studies of the impact of this screening are currently being carried out. Bilecik is located in the intersection of the Marmara, and the Central Anatolia, Black Sea, and Aegean regions. According to the Turkish Statistical Institute (TUIK), the current population of Bilecik is 218, 297. The aim of the present study was to analyse the hemoglobinopathy test results retrospectively in individuals who applied to the hemoglobinopathy diagnosis centre in Bilecik.

Material and Method

Individuals who applied for pre-marriage counselling and general screening at the Bilecik Hemoglobinopathy Diagnostic Centre from January 2006 to April 2017 were included in this study. The hemoglobinopathy test results were retrospectively analysed. The hematological parameters of EDTA blood samples and percentage values of HbA0, A1C, A2, F, C, D, E, and S were determined by using High Performance Liquid Chromatography Bio Rad D-10 (Hercules, USA). The samples with HbA2 values of 3.5% were accepted as indicating traits of beta-thalassemia. The detected HbS (S-Window), HbD (D-Window), and HbC (C-Window) fractions with HbA0 were evaluated using the same method. The statistical analyses were performed with the SPSS 23.0 for Windows (SPSS Inc., USA) program.

Results

A total of 2,676 individuals applied for hemoglobinopathy screening to the Bilecik Hemoglobinopathy Diagnostic Centre from January 2006 to April 2017. The number of beta-thalassemia traits identified was 1,317 (5.56%) in 23,676 premarital screening and general screening individuals. Of these patients, 1,177 had an HbA2 trait level over 3.5%. In 324 (1.3%) patients, elevated HbF levels were over 2%. Both traits were seen together in 184 patients. Beta-thalassemia was detected in only 15 couples of patients who were traits. The ratios of the traits identified, by years, are shown in Table 1 and Figures 1 and 2.

HbD trait (0.06%) was detected in 15 patients (0.06%) and HbS trait was detected in 2 patients (0.01%). The HbE trait was not found (Figure 3).

Table 1. Carrier rates by year

YEAR	Elevated HbF levels (%)	HbA ₂ carrier (%)*	Total Carrier Ratio
2006	1,2	2,9	3,63
2007	2,69	8,3	9,02
2008	1,54	4,7	4,8
2009	1,64	4,15	4,44
2010	1,15	6,8	6,85
2011	1,27	8,82	9,24
2012	1,49	4,91	5,73
2013	0,99	3,68	4,12
2014	1,5	3,29	4,07
2015	1,75	4,31	5,4
2016	0,97	4,26	4,9
2017	0,86	8,6	9,1
Total	1,37	4,6	5,5

*: For HbA2 carrier, HbA2 > 3.5% was accepted.

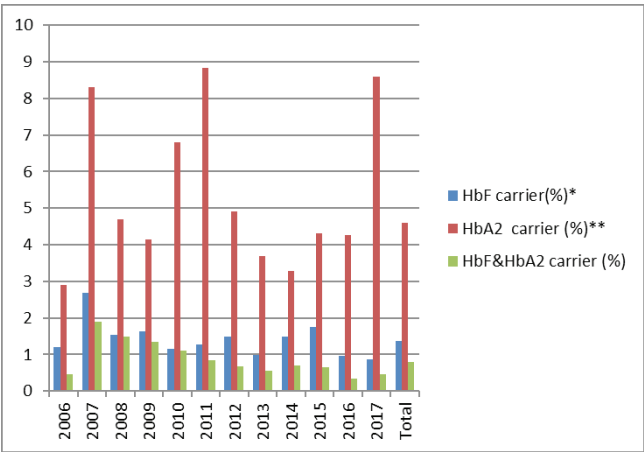


Figure 1. Hemoglobinopathy Carrier Rates by Year

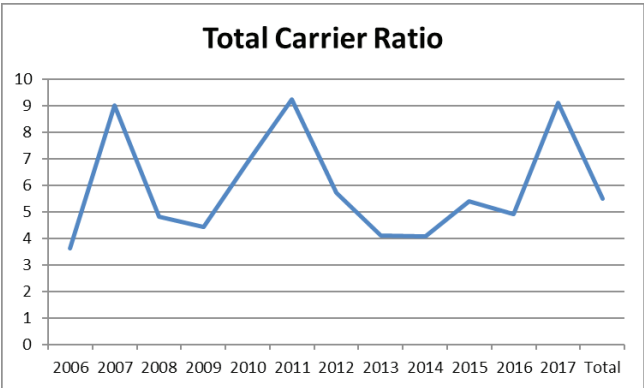


Figure 2. Total Carrier Rates by Year

Discussion

Hemoglobinopathies are an important health problem especially in developing countries [1]. In Turkey, significant reductions in the frequency of beta-thalassemia and sickle cell anaemia (SCA) have been achieved with screening and informing studies in areas of high hemoglobinopathies [2,3,5].

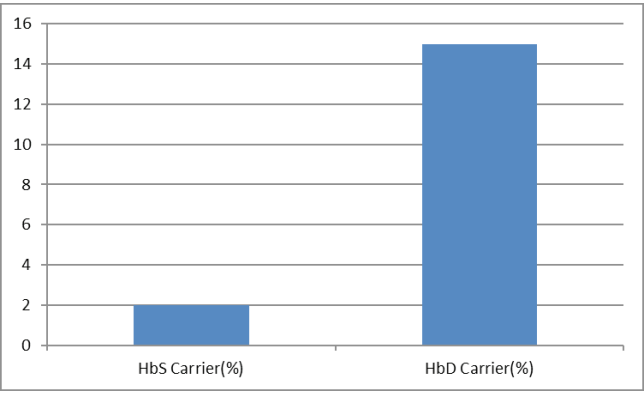


Figure 3. HbS and HbD Carrier Ratios

The overall beta-thalassemia carrier rate is reported as 2.1% in Turkey. This carrier frequency varies between 0.7% and 13% in different cities of Turkey. For instance, the rate is 1.7% in Bursa, which is very close to Bilecik, while it is 4.5% in Istanbul. The beta-thalassemia carrier rate ranges from 0.68% to 4.91% in studies conducted in different regions of our country. The vast majority of studies are based on HbA2 ratios. In our study the HbA2 ratio was 4.6%; this result is similar to the work done in the province of Istanbul and in the Mediterranean region, where the rate was found to be higher than in studies completed in the provinces of Kocaeli and Bursa [5-14]. This is thought to be due to the recent increased immigration to Bilecik, which is at the intersection of the four regions, and the increase of foreign asylum-seekers.

The most common forms of abnormal hemoglobins that affect a large number of people are HbS, HbD, HbE, and HbC. In our study, the HbS trait (0.01%) was observed. One of the most common abnormal hemoglobins in Turkey, HbS causes sickle cell anaemia (SCA). Recent studies have shown a low prevalence of sickle cell anaemia carriers. HbS (Homozygote - HbSS) was detected in 286 fetuses in a study of 1,575 people in 2008 covering the southern region of Turkey [15]. In another study conducted in Kahramanmaraş, HbS carrier was reported as 0.54%. In the study conducted in 2009 in Kocaeli province, HbS carrier was found to be 0.05% [12]. The striking difference between studies is thought to be because the distribution of abnormal hemoglobin varies considerably among geographic areas and racial groups.

Another finding of our study was an HbD carrier rate of 0.06%. The prevalence of HbD in the study conducted in Kahramanmaraş was found to be 0.28% [16]. The appearance of HbD at different ratios suggests a polymorphism [17,18]. It is also believed that the limits of detection of certain variants of the devices differ and that many variants cannot be detected because each method does not show sufficient sensitivity.

Scientific Responsibility Statement

The authors declare that they are responsible for the article's scientific content including study design, data collection, analysis and interpretation, writing, some of the main line, or all of the preparation and scientific review of the contents and approval of the final version of the article.

Animal and human rights statement

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. No animal or human studies were carried out by the authors for this article.

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Conflict of interest

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